GROWING FAMILIES™...

A Newsletter For Those Who Care For Childbearing Families and Neonates

Volume 9, No. 2, February, 2005 ©METABOLIC SCREENING OF THE NEWBORN

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Metabolic screening of the newborn is diagnostic testing performed shortly after birth, designed to allow for early identification and treatment of selected disorders. These conditions include inborn errors of metabolism and/or endocrine disorders that may, if undetected, cause irreversible damage or death to the affected infant.

A simple test, however, can change the course of life for these affected children. In many cases, simple dietary changes or supplements can prevent the severe neurologic damage, disability and death that might otherwise occur. Healthcare providers working with childbearing families are in a key position to promote the health of infants via this vital screening process, and to educate parents regarding the importance of newborn screening.

This newsletter will discuss the types of screening performed in the US, new technologies in use, and how the process of newborn screening is typically carried out. The role of the healthcare provider in assisting with this process and in educating parents will also be examined.

NEWBORN SCREENING IN THE USA

Routine newborn screening, on a large scale, was first conducted in the US in the early 1960s. A test was developed that could provide early screening for phenylketonuria (PKU), a metabolic disorder that, without appropriate treatment, can cause mental retardation. This simple screening test has allowed thousands of children to enjoy normal, healthy lives, and opened the door to the possibility of more widespread screening for many other disorders.

According to data published by the National Newborn Screening and Genetics Resource Center, newborn screening is currently required by every state. More than 4 million newborns are currently screened each year in the US. There is wide variation between states, however, in which tests are required. There is current scientific knowledge and ability to screen for over 50 known metabolic and/or endocrine disorders in the newborn. Most states, however, screen for only a small percentage of those. All states currently screen for PKU, congenital hypothyroidism, and galactosemia. Most also screen for disorders such as:

- sickle cell disease
- maple syrup urine disease
- congenital adrenal hyperplasia
- biotinidase deficiency
- homocystinuria

There are no federal regulations in place requiring specific newborn screening tests. Therefore, the type of screening conducted is highly variable from state to state. State mandated screening may range from just three disorders, to widely expanded screening, including diseases such as cystic fibrosis, toxoplasmosis, and HIV. The March of Dimes currently recommends screening for at least 29 disorders, and urges states to consider additional testing for another 25 conditions..

COMMON DISORDERS

The metabolic and endocrine disorders most commonly screened for in the US include the following:

Phenylketonuria: Incidence 1/14,000 births. PKU is caused by the lack of an enzyme to metabolize the amino acid phenylalanine. Found in milk, meats and other protein foods, phenylalanine then builds up in the blood, causing neurologic damage. Special formula and a low-phenylalanine diet can completely prevent this damage.

Congenital hypothyroidism: Incidence 1/4,000 births. Hypothyroidism in the newborn, lack of thyroid hormone, may result from faulty development of the thyroid gland or enzyme deficiencies. This is the most common disorder detected by newborn screening. Without adequate thyroid hormone, severe mental retardation and stunting of growth occurs. These effects can be prevented by early replacement of oral thyroid hormone.

Galactosemia: Incidence 1/50,000 births. This disorder results from lack of an enzyme that metabolizes galactose, a sugar found mainly in milk products. When galactose builds up, mental retardation, stunted growth, cataracts, and damage to the liver and kidneys may occur. The majority of affected infants will die without treatment. To prevent these effects, persons with galactosemia must avoid all sources of galactose and lactose (which is broken down to galactose) for life.

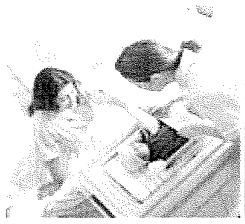
NEWBORN SCREENING TECHNOLOGY

Newborn screening became feasible in the 1960's largely due to the work of Dr. Robert Guthrie, the father of a child with mental retardation. Dr. Guthrie developed a test to detect PKU using blood collected on a spot of filter paper and allowed to air-dry. He then worked tirelessly to promote widespread newborn screening for PKU.

The same blood collection method, called the Guthrie spot test, is in use today for newborn screening. One aspect of newborn screening that has changed a great deal from Dr. Guthrie's time, however, is the way in which blood samples can now be analyzed. Many states are now using tandem mass spectrometry (MS/MS) technology to analyze blood samples for newborn screening. MS/MS allows the detection of multiple disorders from a single blood sample, making screening for a variety of conditions efficient and feasible.

ROLE OF THE HEALTHCARE PROFESSIONAL

It is critically important that healthcare professionals who work with childbearing families know what disorders are included in their state's newborn screening program.



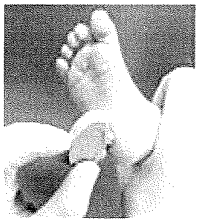
This information should be provided to parents in both written and verbal form. In addition, some parents are now also choosing the option of expanded screening through independent laboratories, and these options should also be presented.

State requirements must be followed regarding timing of specimen collection. For example, specimen collection may be required within 72 hours of birth, with testing preferably after the first 24 hours of life. Testing after 24 hours increases the accuracy of those tests influenced by milk intake, such as PKU, maple syrup urine disease and homocystinuria.

A second screen should be performed at 1-2 weeks of age when the initial testing is done before 24 hours, or when required by state law. Regardless of the newborn's age, blood collection should be done before the newborn is discharged to home. If a blood transfusion or IV antibiotic therapy is necessary, collection should occur before such interventions are started. They may falsely affect results.

Before specimen collection, all requested information on the newborn screening card must be filled out so that it is complete and legible. This card also contains the filter paper circles in which the blood is collected, and these should not be touched with gloved or ungloved hands.

In most cases, blood is taken from the newborn's heel after puncture with a lancet or other device. As always, standard precautions, including use of gloves, should be used during this procedure. Since the quality of the specimen depends in large part on the blood collection technique, it is very important that collection instructions be followed precisely to obtain accurate results.



To promote good blood flow from the puncture site, the newborn's heel should be prepared by wrapping in a towel or diaper soaked in warm water. The newborn's head should be higher than the feet, either by being held upright or by raising the head of the crib. The foot and leg should not be squeezed or "milked" during collection, as this may release excess tissue fluid that dilutes the sample. The filter paper should be lightly touched to large drops of blood forming on the heel, until the paper is soaked through to the back. Since accuracy of the screen is dependent on the amount of blood filling each circle, the circles must be completely filled, and from one side only. Using multiple drops to fill a circle should be avoided, as this produces a layered or clotting effect that may invalidate results. After all circles are completely filled, the specimen must then be allowed to air-dry, avoiding exposure to direct sun or heat, before being sent to the laboratory.

Let the parents know that the specimen has been collected, and encourage them to ask about the results at the baby's first check-up. Emphasize that, if any results come back positive, additional testing will be necessary to make a diagnosis—these tests are for screening purposes only. Also, let the parents know if a second screening is required.

The healthcare professional's careful attention to the newborn screening process helps to promote early identification of newborns at risk for metabolic and other disorders.

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